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(21) International Application Number: PCT/US96/19046 (22) International Filing Date: 15 November 1996 (15.11.96) (30) Priority Data: 08/559,303 15 November 1995 (15.11.95) US (71) Applicant: NEW YORK BLOOD CENTER, INC. [US/US]; 310 East 67th Street, New York, NY 10021-6295 (US). (72) Inventors: ELLIS, Nathan; 256 West 10th Street, New York, NY 10014 (US). GERMAN, James; 270 Riverside Drive, New York, NY 10025 (US). GRODEN, Joanna; 3513 Clarkwood Place, Cincinnati, OH 45208 (US). (74) Agents: BOGOSIAN, Elizabeth, A. et al.; 90 Park Avenue, New York, NY 10016 (US).		(81) Designated States: CA, IL, JP, European patent (AT, BE, CH, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE). Published <i>With international search report. Before the expiration of the time limit for amending the claims and to be republished in the event of the receipt of amendments.</i>
(54) Title: METHODS FOR DIAGNOSIS AND TREATMENT OF BLOOM'S SYNDROME (57) Abstract The present invention provides a method for diagnosing BS as well as determining whether a subject is a carrier of a mutated <i>BLM</i> gene. The present invention also provides one or more single-stranded nucleic acid probes and antibodies which may be formulated in kits, and used for diagnosing BS or determining whether a subject is a carrier of a mutated <i>BLM</i> gene. In addition, the present invention provides a method for treating or preventing the onset of BS in a subject in need of such treatment or prevention, as well as vectors and stem cells useful for such treatment or prevention. The present invention also provides a purified and isolated nucleic acid encoding an enzymatically active <i>BLM</i> protein, a vector comprising this nucleic acid, a cell stably transformed with this vector, as well as a method for producing recombinant, enzymatically active <i>BLM</i> protein. A purified, enzymatically active <i>BLM</i> protein is also provided by the present invention. Finally, the present invention provides a vector, an embryonic stem cell, and a non-human, transgenic animal, each of which comprises a mutated <i>BLM</i> gene, as well as a method for producing the non-human, transgenic animal.		